Case Report

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A Case Report of Diaper Staining Since Birth: Rare Metabolic Disorder Alkaptonuri

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Abstract

Alkaptonuria is a rare metabolic autosomal recessive disorder caused by deficiency of homogentisic acid oxidase [1]. A three old female child presented with history of staining of diaper since birth and during treatment underwent battery of scanning and investigations. Diagnosis was suspected when urine turned black when kept in a sterile container. Her urine examination showed homogentisic acid. Patient was diagnosed alkaptonuria.

Keywords: Genetic Disorder; Alkaptonuria.

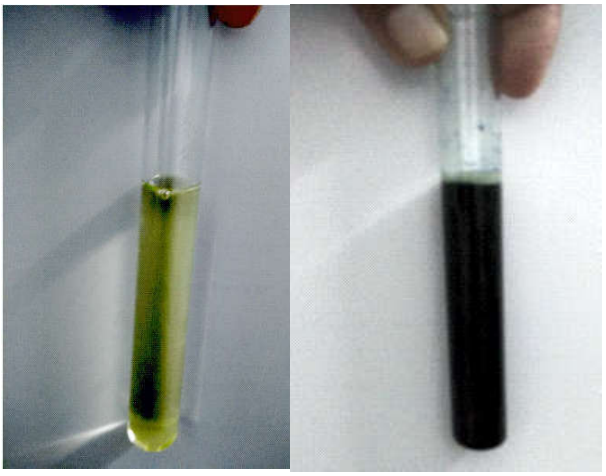
Introduction

Alkaptonuria is a rare disorder of tyrosine metabolism due to deficiency of homogentisic oxidase [1]. In alkaptonuria large amount of homogentisic acid are formed which are excreted in urine or deposited in tissues [2,3,4]. The only clinical signs in children is blackening of urine on standing caused by oxidation and polymerization of homogentisic acid. Metabolites of homogentisic acid are deposited in tissues leading to ochronosis in adulthood. It is seen clinically as dark spots in sclera in eyes and ear cartilage. Another manifestation of deposition is cardiac valvulitis and arthritis leading to destruction of joints [5,6]. We report a child who attended pediatric opd with complaints of dark staining of diapers.

Case Report

A three year old female child of a third degree consanguineous couple presented to OPD with history of staining of diaper since birth. This was noticed by mother in neonatal period during which parents did not consulted to any doctor initially but later child underwent lots of investigation, all were normal. Child had no other complaints. Child was well built and nourished. On examination no abnormality detected. Child was noticed during voiding of urine. Initially urine appeared normal but after 5-10 minutes of standing it turned black. Routine laboratory investigation were within normal limits. Skeletal X-rays were normal.

Urine Gas chromatography/ Mass Spectrometry showed a massive amount of homogentisic acid. Child was started on phenylalanine and tyrosine restricted diet and vitamin C. child is under regular follow up.



Just after voiding after 10 minutes

Discussion

In my case child's urine was normal in appearance while voiding but on standing for sometime it turns dark in colour. This darkening of urine is caused by oxidation and polymerization of homogentisic acid and the action of the metabolite is enhanced in alkaline environment. Sometimes diagnosis may be delayed up to adulthood, when arthritis and ochronosis occurs because acidic urine may not become dark even after many hours of standing [1]. The diagnosis of the disease is confirmed by measurement of homogentisic acid in urine or by the high pressure liquid chromatography method for quantification of homogentisic acid and its derivative benzoquinone acetic acid [2]. Excretion of homogentisic acid in urine is usually 4-6g daily and its measurement is also used for therapeutic monitoring.

Sometimes pediatric alkaptonuria may be asymptomatic but in our case blackish discoloration of diaper was discovered by mother accidentally. Pigmentation of sclera or the cartilage of the ear usually appears in adulthood [1]. Pigmentation may also be seen in the teeth [4], buccal mucosa, skin giving these areas a dusty color, which is due to slow accumulation of the black polymer of homogentisic acid in the cartilage and other mesenchymal tissues [3]. A case was reported in which B/L spontaneous rupture of quadriceps tendon was initial presentation [5].

Arthritis occurs in almost all patients with advancing age and it is the only disabling effect of the condition. It appears early in the large weight bearing joints like hip, spine and knees. Cardiac involvement includes high incidence of heart diseases commonly valvulitis involving aortal and mitral valves [3,7]. Ischaemic heart disease with ultimate myocardial infarction is a common cause of death.

Treatment of alkaptonuria patients is a challenge. No treatments have been completely successful. Dietry restriction on tyrosine and phenylalanine aminoacids reduces excretion of homogentisic acid. Ascorbic acid prevents the effects of this metabolites on joints [8]. Nitisinone has been proposed as potential theraphy because it inhibits the enzyme that produces homogentisic acid but at present it is still under trial [6].

Conclusion

Any complaint of discoloration of urine should not be ignored especially if routine investigations are normal.

Conflicts of Interest: The author have no conflicts of interest

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